SickKids Genome Diagnostics

IDENTITY TESTING: TISSUE MATCHING

Individuals can be distinguished from one another by DNA fingerprinting, which compares variable DNA markers in different regions of the genome. Each person has his or her own pattern of DNA markers - a DNA 'fingerprint'. PCR amplification followed by capillary electrophoresis allows comparison of these markers between different individuals.

DNA fingerprinting can also be used to evaluate the purity of tissue samples obtained for other kinds of tests. Maternal cell contamination (MCC) of fetal cell samples obtained for prenatal analysis can be assessed in this way, and DNA fingerprinting can also be used to distinguish between the X and Y chromosomes for fetal sex determination.

TEST METHODS

Identity testing is performed by studying 15 DNA microsatellite markers and a sex-differentiating marker (D5S818, vWA, D13S317, THO1, D7S820, TPOX, CSF1PO, D8S1179. D21S11, D3S1358. D16S539, D2S1338, D19S433, D18S51, FGA and AMELX/Y). amplification these PCR of targeted regions followed by electrophoresis allows capillary comparison of these markers in the samples provided.

The test sensitivity level is 5% for detection of mixed samples.

TISSUE MATCHING

Samples from the same person taken at different times are compared to confirm that the samples have not been mixed up. The samples are confirmed to be from the same source when they match at all of the DNA markers used in the analysis.

DNA fingerprinting does not map the entire genome of the samples, thus there exists a small probability of obtaining a false match. A risk estimate of this occurrence is provided with each report.

Contamination of samples are detectable by molecular analysis at levels to 5%. The additional, disease-specific analysis of samples contaminated at ratios lower than this may be adversely affected or inconclusive.

For More Information

Understanding Gene Testing: <u>http://www.accessexcellence.</u> org/AE/AEPC/NIH/index.html

Waye JS (1993) Forensic identity testing using highly polymorphic DNA markers: current status and emerging technologies. *Transfusion Medicine Review* 7: 193-205.

To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at <u>www.cagc-accg.ca</u> or the National Society of Genetic Counsellors website at <u>www.nsgc.org</u>

SickKids

1. This test was developed anditsperformance characteristics validated by the Genome Diagnostics Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drua Administration. The FDA has determined t hat su ch clearance or approval is not necessary. This test is used for clinical purposes.